TEST ID: 63129
ALPORT (COLLAGEN IV ALPHA 5 AND ALPHA 2) IMMUNOFLUORESCENT STAIN, RENAL

USEFUL FOR
Assisting in the diagnosis of hereditary nephritis (Alport syndrome)

CLINICAL INFORMATION
Alport syndrome is a hereditary disease of basement membrane collagen type IV. Mutations in collagen IV alpha genes cause characteristic abnormal immunofluorescence staining patterns within the glomerular basement membrane. Alport syndrome is characterized by hematuria, proteinuria, progressive renal failure, and high-tone sensorineural hearing loss.

SPECIMEN REQUIRED
Type: Tissue
Source: Kidney or Skin
Container/Tube: Renal Biopsy Kit (Supply T231), Zeus/Michel’s, Frozen
Specimen Volume: Entire specimen

INTERPRETATION
This test, (when not accompanied by a pathology consultation request) will be reported as: 1) normal pattern, 2) consistent with X-linked hereditary nephritis, or 3) consistent with autosomal hereditary nephritis.

If additional interpretation or analysis is needed, request 70012 / Pathology Consultation along with this test and send the corresponding renal pathology light microscopy and immunofluorescence (IF) slides (or IF images on a CD), electron microscopy images (prints or CD), and the pathology report.

MOBILE APPS FROM MAYO MEDICAL LABORATORIES
Lab Catalog for iPad and Lab Reference for iPhone and iPod Touch
Requires iOS 5.1+

REFERENCE VALUES
Detailed on the back side of this sheet

ANALYTIC TIME
Turnaround time is 1 working day (from receipt)

CONTENT AND VALUES SUBJECT TO CHANGE. SEE THE MAYO MEDICAL LABORATORIES TEST CATALOG FOR CURRENT INFORMATION.
REFERENCE VALUES

Reporting of immunofluorescent (IF) double staining for alpha 2 and alpha 5 chains of type IV collagen on kidney biopsies:

- Normal pattern of staining (ie, preserved linear alpha 5 staining of glomerular basement membranes, Bowman capsule, and distal tubular basement membranes). This pattern of staining is seen in normal individuals and patients with thin glomerular basement membrane disease but does not exclude the diagnosis of hereditary nephritis/Alport syndrome.

- Consistent with X-linked hereditary nephritis (Alport syndrome). There is global or segmental loss of alpha 5 staining of glomerular basement membranes, Bowman capsule, and distal tubular basement membranes. This pattern of loss of staining is usually due to mutations in the \( \text{COL4A5} \) gene on the X chromosome.

- Consistent with autosomal hereditary nephritis (Alport syndrome). There is global or segmental loss of alpha 5 staining of glomerular basement membranes but preserved alpha 5 staining of Bowman capsule and distal tubular basement membranes. This pattern of loss of staining is usually due to mutations in the \( \text{COL4A3} \) or \( \text{COL4A4} \) genes on chromosome 2.

- No interpretation can be reported if the specimen contains no intact glomeruli.

Reporting of IF double staining for alpha 2 and alpha 5 chains of type IV collagen on skin biopsies:

- Normal pattern of staining (ie, preserved linear alpha 5 staining of epidermal basement membranes). This pattern of staining is seen in normal individuals and patients with thin glomerular basement membrane disease but does not exclude the diagnosis of hereditary nephritis/Alport syndrome.

- Consistent with X-linked hereditary nephritis (Alport syndrome): There is global or segmental loss of alpha 5 staining of epidermal basement membranes. This pattern of loss of staining is usually due to mutations in the \( \text{COL4A5} \) gene on the X chromosome.

- No interpretation can be reported if the biopsy contains no epidermis.

Notes:

- Approximately one-third of patients with established hereditary nephritis based on typical ultrastructural findings and family history show loss of glomerular basement membrane or epidermal basement membrane staining for the alpha 5 chain of type IV collagen. Therefore, a normal staining pattern does not exclude the diagnosis of hereditary nephritis.

- In patients with hereditary nephritis, preserved alpha 5 staining indicates small mutations (eg, missense, splice site) and is generally associated with a better renal outcome, while loss of alpha 5 staining indicates larger mutations (eg, deletion, nonsense, frame-shift) and a worse renal outcome.

- Because alpha 3 and alpha 4 chains of type IV collagen are not expressed in the epidermal basement membranes, patients with autosomal hereditary nephritis have preserved staining for alpha 5 on epidermal basement membranes and, therefore, skin biopsy cannot exclude autosomal hereditary nephritis.